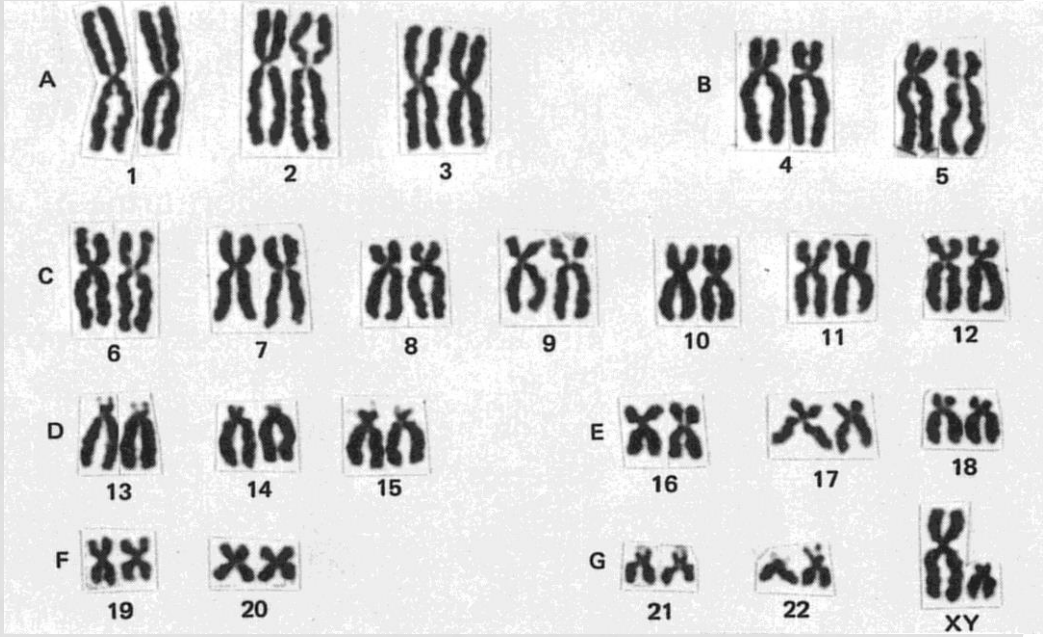
THE HUMAN GENOME

CH 14

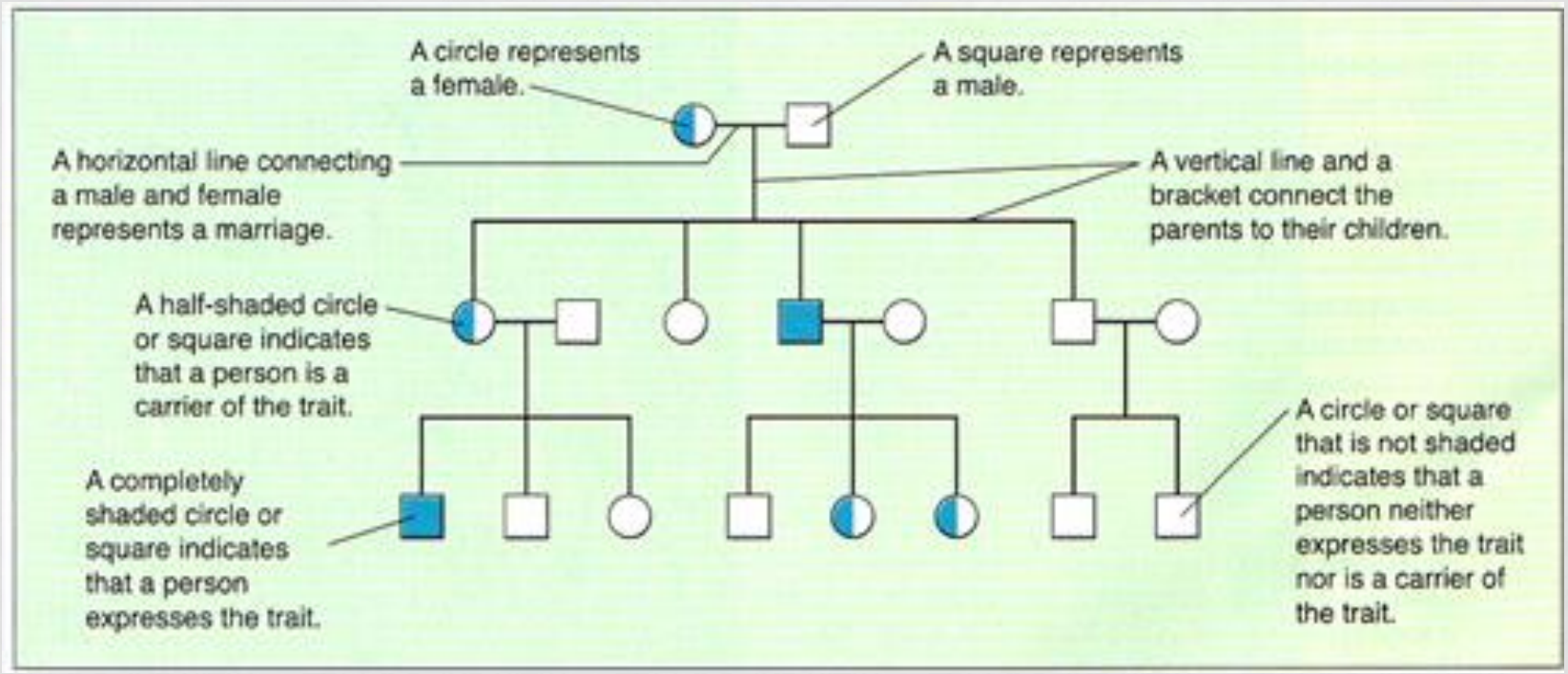
14-1 HUMAN HEREDITY

- Karyotypes are pictures of human chromosomes
- 1N (Haploid) + 1N egg=diploid zygote
- 46 chromosomes, 23 different ones
- 2 are sex chromosomes
- Females XX, Males XY

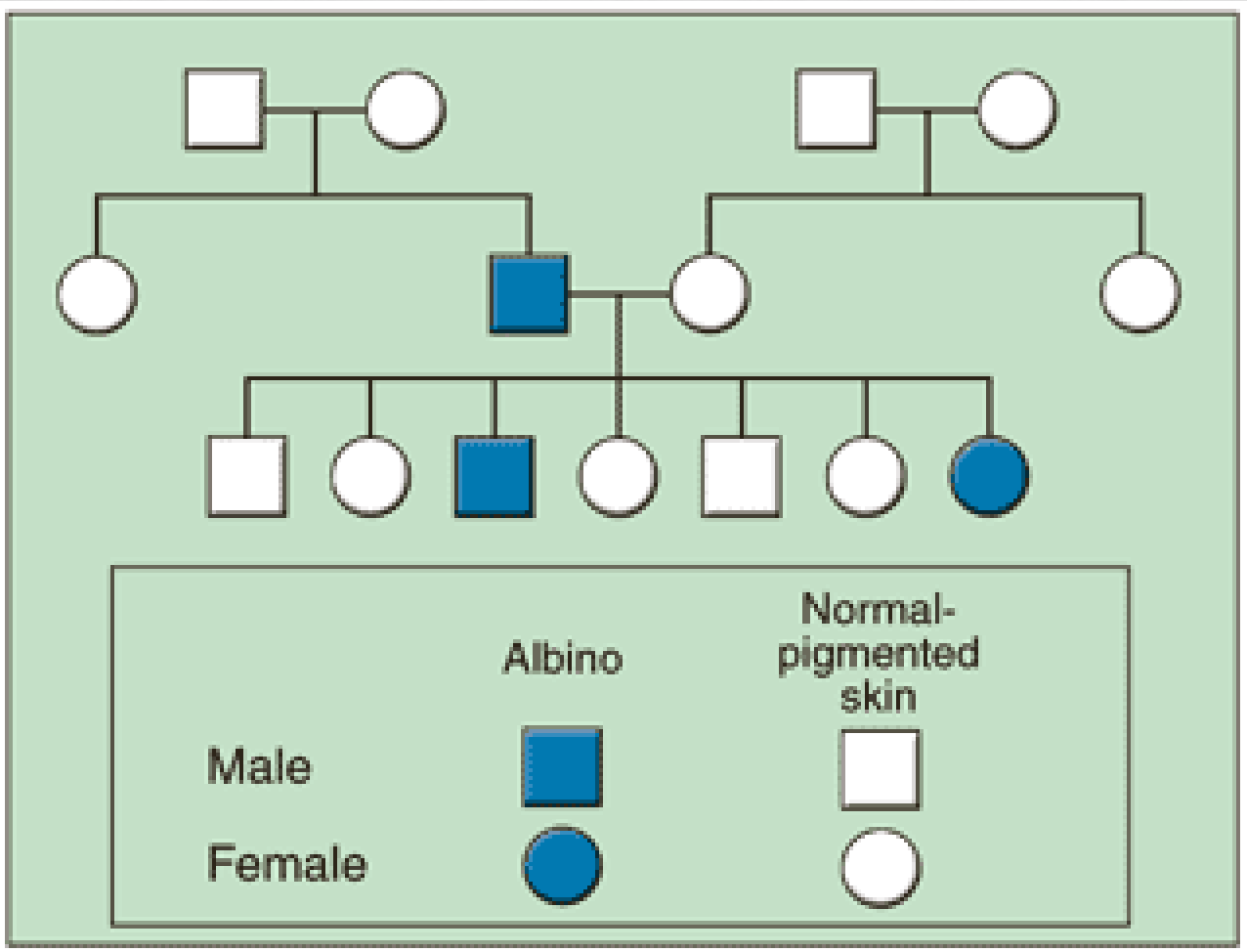


14-1 HUMAN HEREDITY

- Pedigrees show how traits are passed from one generation to the next



14-1 HUMAN HEREDITY

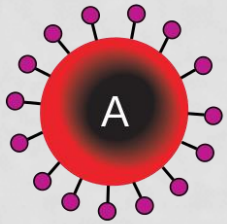
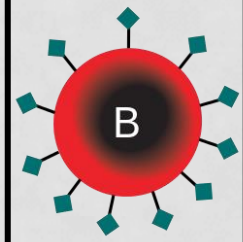
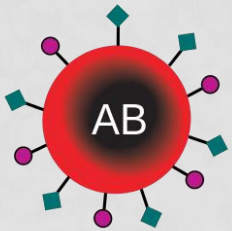
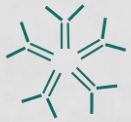
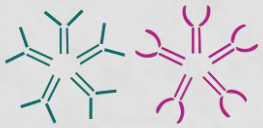


14-1 HUMAN HEREDITY

- Human Genome has about 30,000 genes

14-1 HUMAN HEREDITY

- Blood Group Genes-ABO and Rh groups
- Rh-2 alleles, +/- (+ is dominant)
- ABO-three alleles

	Group A	Group B	Group AB	Group O
Red blood cell type				
Antibodies in Plasma	 Anti-B	 Anti-A	None	 Anti-A and Anti-B
Antigens in Red Blood Cell	 A antigen	 B antigen	 A and B antigens	None

14-1 HUMAN HEREDITY

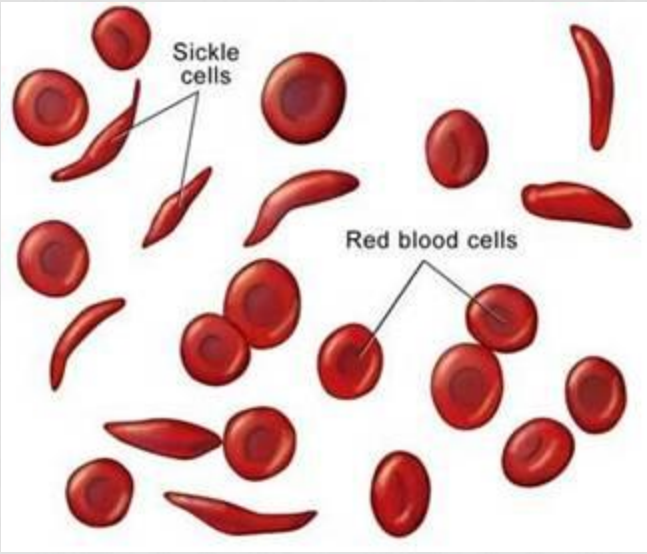
- Recessive alleles-only see phenotype when the normal dominant allele fails
- PKU-lack enzyme to break down amino acid phenylalanine
- Causes severe mental retardation
- Individuals need a diet low in phenylalanine
- Tay-Sachs disease affects Jewish families in central and eastern Europe-causes nervous system breakdown in first 2 years of life

Autosomal Disorders

Some Autosomal Disorders in Humans		
Type of Disorder	**Disorder**	**Major Symptoms**
Disorders caused by recessive alleles	Albinism	Lack of pigment in skin, hair, and eyes
	Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in childhood unless treated
	Galactosemia	Accumulation of galactose (a sugar) in tissues; mental retardation; eye and liver damage
	Phenylketonuria (PKU)	Accumulation of phenylalanine in tissues; lack of normal skin pigment; mental retardation
	Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in early childhood
Disorders caused by dominant alleles	Achondroplasia	Dwarfism (one form)
	Huntington's disease	Mental deterioration and uncontrollable movements; appears in middle age
	Hypercholesterolemia	Excess cholesterol in blood; heart disease
Disorders caused by codominant alleles	Sickle cell disease	Sickled red blood cells; damage to many tissues

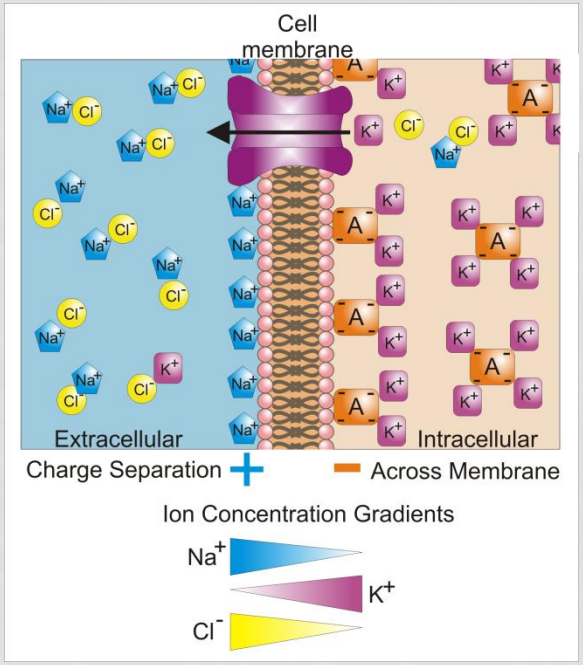
14-1 HUMAN HEREDITY

- Diseases can also be caused by dominant alleles- achondroplasia, Huntingtons Disease
- Sickle cell anemia caused by a co-dominant allele- but cell shape protects against Malaria infection



14-1 HUMAN HEREDITY

- Changes in DNA lead to changes in protein structure and function-that is what causes the disease
- Cystic fibrosis is caused by a mutation that causes a small change in a protein
- that allows Cl- ions to
- pass through
- membranes. Causes
- tissues throughout the
- body to malfunction

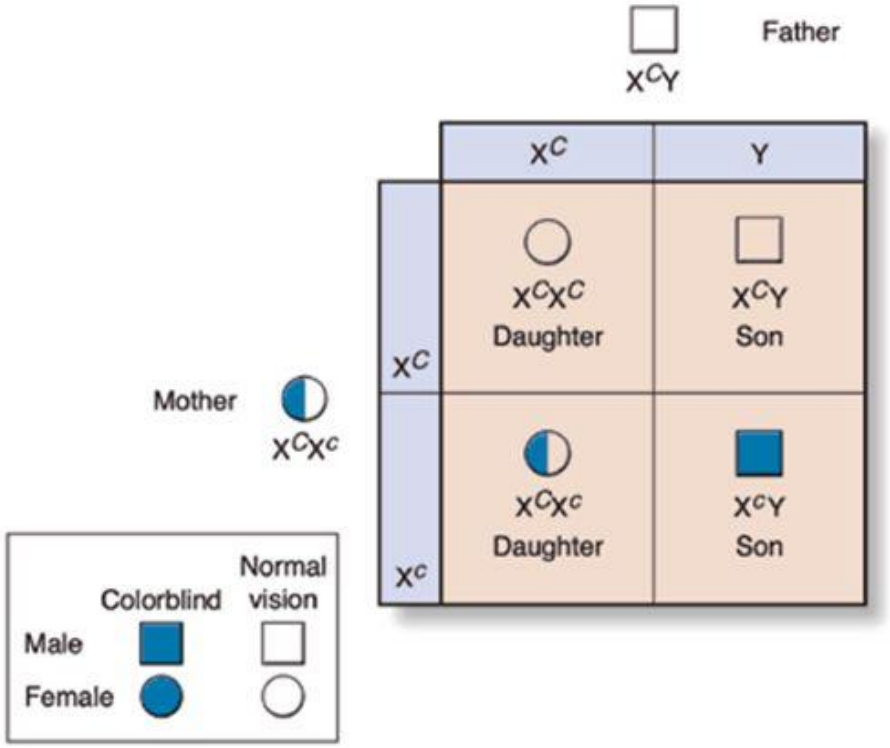


14-2 HUMAN CHROMOSOMES

- 6 billion bases in diploid cell
- If a book, 4 million pages long
- Smallest chromosomes (21&22) have 30-40K bases
- Sex linked genes located on X&Y chromosomes
- More than 100 sex-linked genes on X&Y chromosomes
- All x-linked alleles are expressed in males; in females they need 2 copies of the allele to be expressed; sex-linked disorders show up more in males than in females
- Hemophilia and Duchenne Muscular dystrophy are x-linked diseases

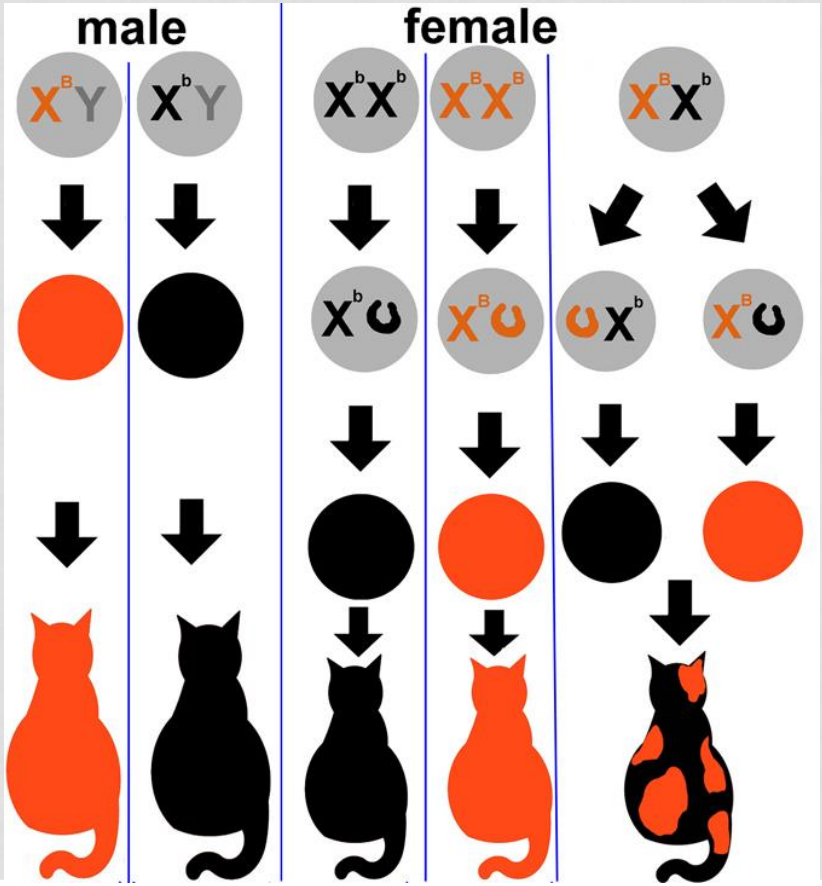
Sex-Linked Genes

- **Males have just one X chromosome. Thus, all X-linked alleles are expressed in males, even if they are recessive.**



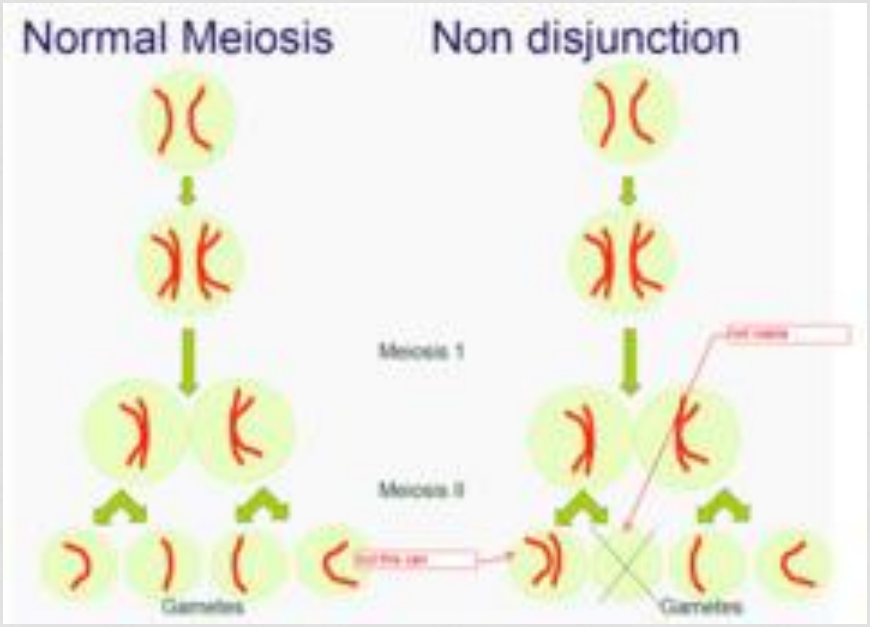
14-2 X-CHROMOSOME INACTIVATION

- In females the extra x chromosome is (randomly)turned off, called a Barr body



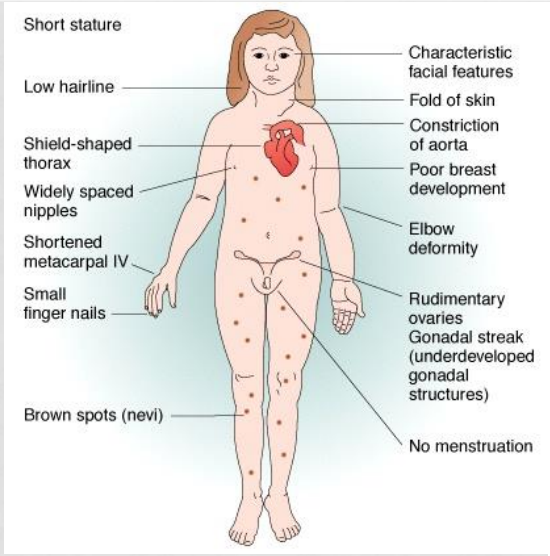
14-2 CHROMOSOMAL DISORDERS

- Nondisjunction during meiosis-when homologous chromosomes do not separate
- Causes abnormal number of chromosomes
- Trisomy 21-Down's Syndrome

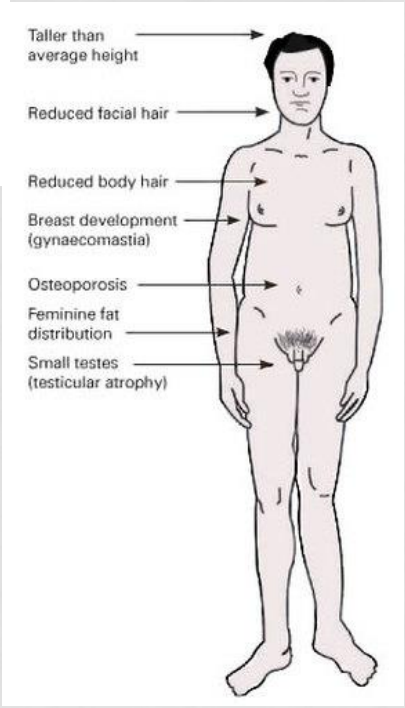


14-2 SEX CHROMOSOME DISORDERS

- Turner's syndrome XO

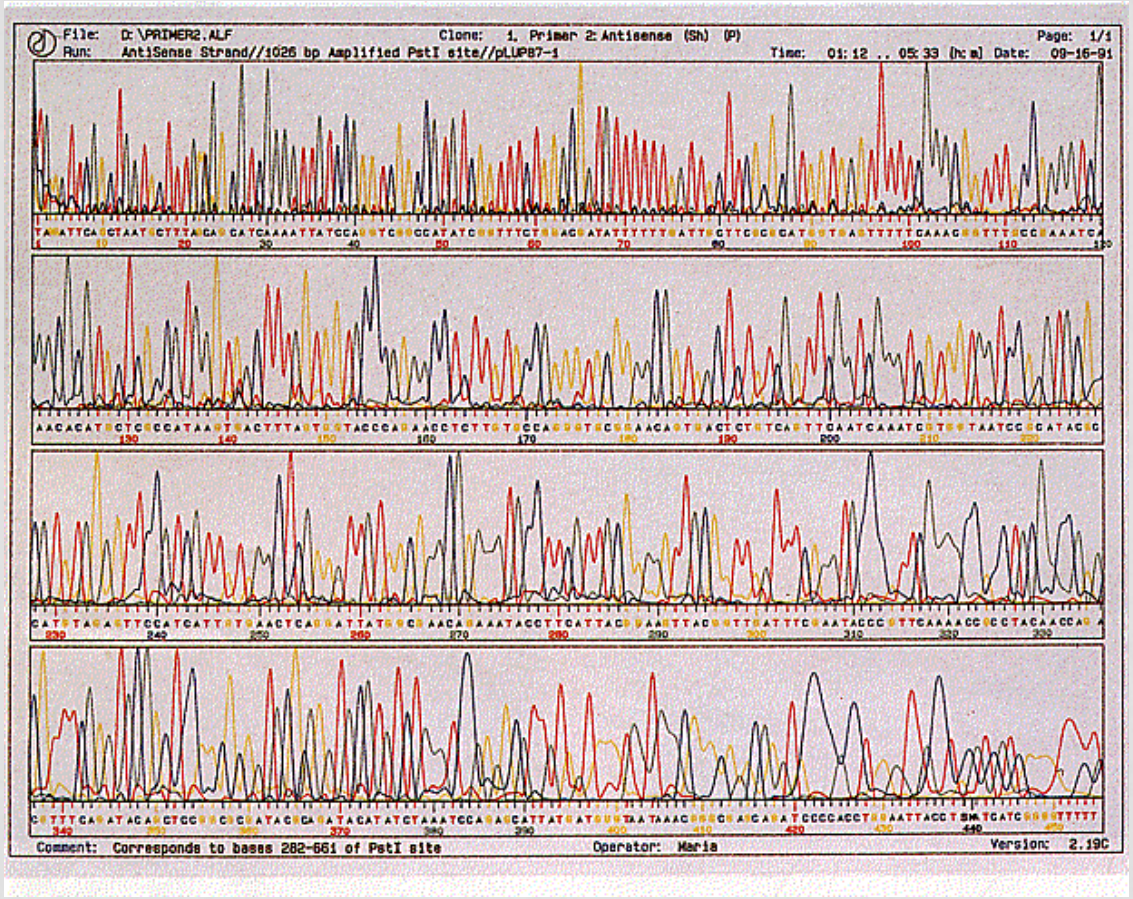


- Klinefelter's Syndrome-XXY



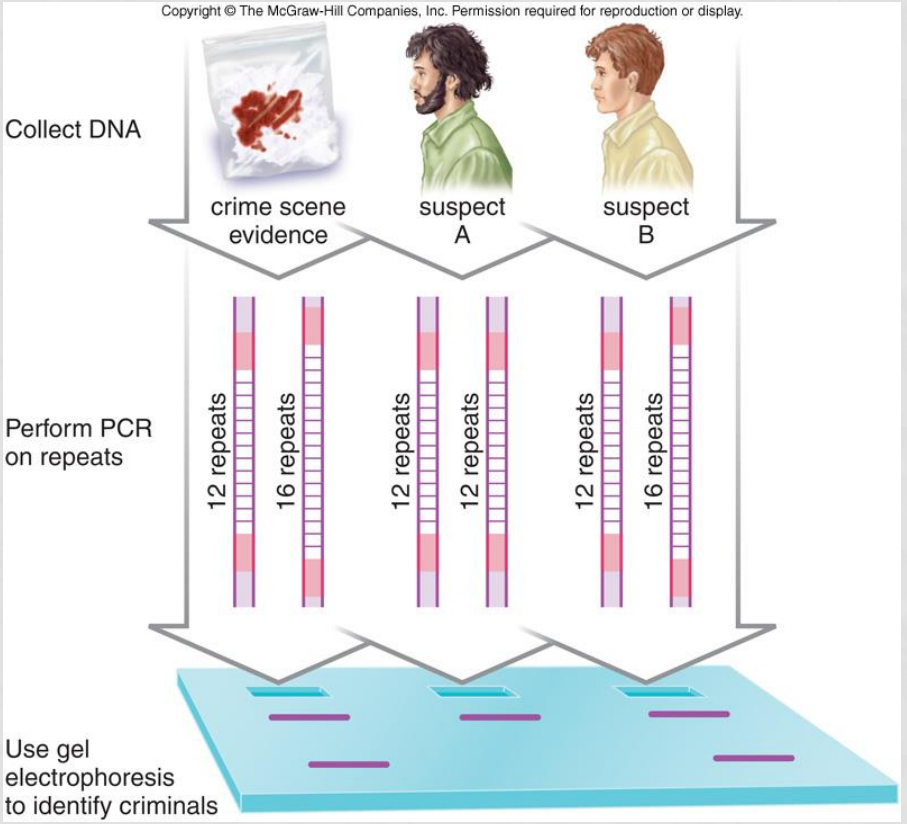
14-3 HUMAN MOLECULAR GENETICS

- Testing for alleles for hundreds of genetic disorders



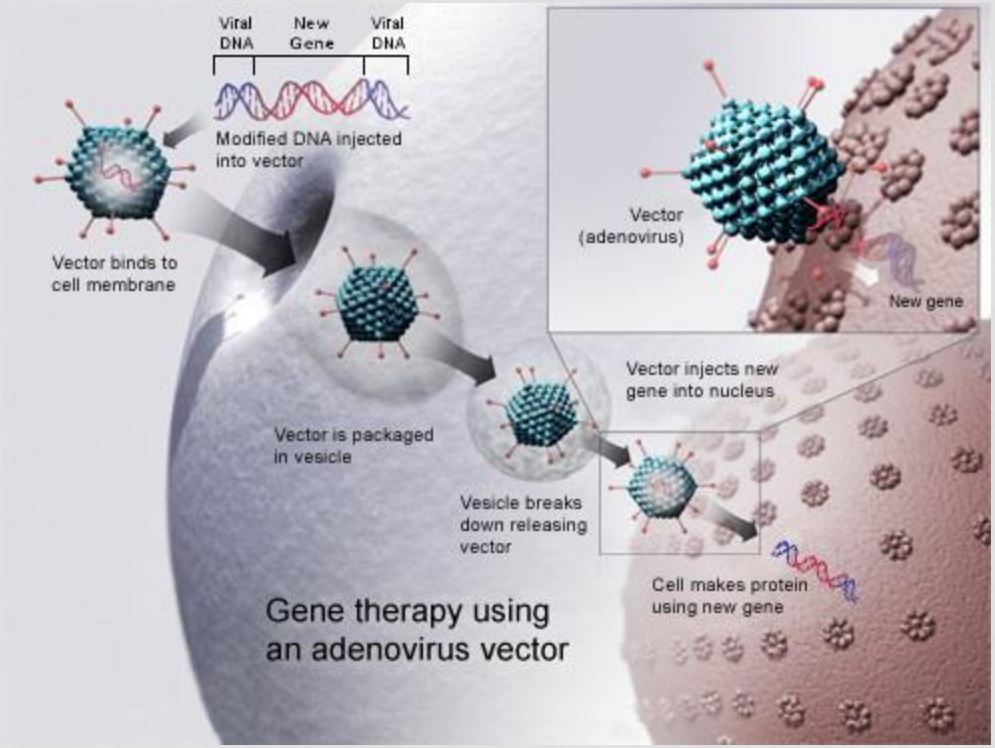
14-3 DNA FINGERPRINTING

- Analyze sections of DNA that are of little importance BUT vary greatly between individuals
- Used for identification



14-3 GENE THERAPY

- An absent or faulty gene is replaced by a normal working gene
- Viruses are used to get the genes inside of cells



14-3 ETHICAL ISSUES IN HUMAN GENETICS

- What will happen to the human species if we can change and design our bodies?
- Need thoughtful and ethical consensus about what is OK or not OK so we can use our new knowledge and tools wisely
- Not just scientists but all of society